



## MUTYH gene

mutY DNA glycosylase

### Normal Function

The *MUTYH* gene provides instructions for making an enzyme called MYH glycosylase, which is involved in the repair of DNA. This enzyme corrects particular mistakes that are made when DNA is copied (DNA replication) in preparation for cell division. DNA is made up of building blocks called nucleotides, each of which has a specific partner. Normally, adenine pairs with thymine (written as A-T) and guanine pairs with cytosine (written as G-C). During normal cellular activities, guanine sometimes becomes altered by oxygen, which causes it to pair with adenine instead of cytosine. MYH glycosylase fixes this mistake so mutations do not accumulate in the DNA and lead to tumor formation. This type of repair is known as base excision repair.

### Health Conditions Related to Genetic Changes

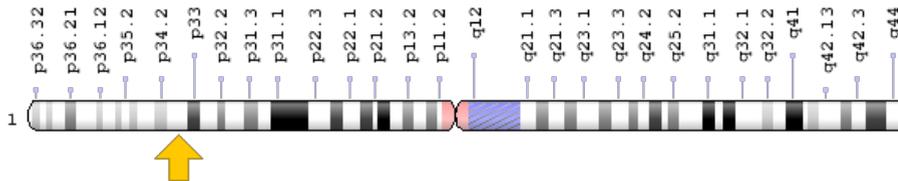
#### familial adenomatous polyposis

Mutations in the *MUTYH* gene cause an autosomal recessive form of familial adenomatous polyposis (also called MYH-associated polyposis). Mutations in this gene affect the ability of cells to correct mistakes made during DNA replication. In individuals who have autosomal recessive familial adenomatous polyposis, both copies of the *MUTYH* gene in each cell are mutated. Most mutations in this gene result in the production of a nonfunctional or low-functioning MYH glycosylase. When base excision repair in the cell is impaired, mutations in other genes build up, leading to cell overgrowth and possibly tumor formation. Two mutations that change the sequence of the building blocks of proteins (amino acids) in MYH glycosylase are common in people of European descent. One mutation replaces the amino acid tyrosine with the amino acid cysteine at position 165 (written as Tyr165Cys or Y165C). The other mutation switches the amino acid glycine with the amino acid aspartic acid at position 382 (written as Gly382Asp or G382D).

## Chromosomal Location

Cytogenetic Location: 1p34.1, which is the short (p) arm of chromosome 1 at position 34.1

Molecular Location: base pairs 45,329,242 to 45,340,925 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- hMYH
- mutY (E. coli) homolog
- mutY homolog
- mutY homolog (E. coli)
- MUTYH\_HUMAN
- MYH

## Additional Information & Resources

### Educational Resources

- American Medical Association and National Coalition for Health Professional Education in Genetics: Understand the Basics of Genetic Testing for Hereditary Colorectal Cancer  
<http://www.nchpeg.org/documents/crc/Basics%20of%20genetic%20testing.pdf>
- Genomes (second edition, 2002): Base excision repairs many types of damaged nucleotide  
<https://www.ncbi.nlm.nih.gov/books/NBK21114/#A8439>

### GeneReviews

- MUTYH-Associated Polyposis  
<https://www.ncbi.nlm.nih.gov/books/NBK107219>

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28MUTYH%5BTIAB%5D%29+OR+%28mutY+homolog%5BTIAB%5D%29%29+OR+%28%28mutY+homolog%5BTIAB%5D%29+OR+%28MYH%5BTIAB%5D%29+OR+%28mutY++homolog%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>

## OMIM

- MutY, E. COLI, HOMOLOG OF  
<http://omim.org/entry/604933>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
<http://atlasgeneticsoncology.org/Genes/MUTYHID41464ch1p34.html>
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=MUTYH%5Bgene%5D>
- HGNC Gene Family: DNA glycosylases  
<http://www.genenames.org/cgi-bin/genefamilies/set/1024>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=7527](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=7527)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/4595>
- UniProt  
<http://www.uniprot.org/uniprot/Q9UIF7>

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